

Complete transposition of great arteries with coarctation of aorta

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SUMMARY Complete transposition of the great arteries with ventricular septal defect and coarctation or atresia of the aortic arch is a relatively rare anatomoclinical entity and in general not well understood. This association accounted for six out of 50 cases (12%) of our clinical experience and six out of 64 cases (9%) of the post-mortem series of complete transposition. Relevant clinical manifestations were cyanosis, early and pronounced congestive heart failure, with a decrease in amplitude of femoral pulses and, in the most severe cases, isolated left ventricular hypertrophy on the electrocardiogram. Inlet or outlet right ventricular obstruction and pulmonary artery dextroposition are the pathological substrates of unequal partitioning of the blood stream in favour of the pulmonary artery and ductus arteriosus, probably accounting for fetal underdevelopment of the aortic arch. The medical and surgical prognosis of these cases is definitely poor.

Obstructive lesions of the aortic arch are usually associated with concordant atrioventricular and ventriculoarterial connection. They may, however, also be present in complete transposition of the great arteries (discordant ventriculoarterial connection with atrioventricular concordance), but they have only sporadically been described in the published reports.¹⁻⁵

It is the purpose of this paper to report our experience with patients affected by complete transposition of the great arteries and coarctation of the aorta. The clinicopathological features of these cases are sufficiently distinctive to indicate that this complex of malformations is a real and not so rare anatomoclinical entity.

Subjects

Among 50 consecutive cases of complete transposition of the great arteries seen at the Pediatric Department of the University of Padua, between June 1975 and June 1980, six (12%) were found to have obstructive lesions of the aortic arch. There were five male and one female patient (Table 1), varying in age from 1 to 55 days (mean 18 days). In three the diagnosis was made by cardiac catheterisation and angiocardiography, five died (cases 1 to 5, Table 1) and came to necropsy, one is still alive (case 6, Table 1). Reviewing our anatomical collection, another specimen (case 7, see Table 3) was found with this association. The incidence in our

necropsy series was 9% (six out of 64 hearts with complete transposition of the great arteries). Only those cases were considered with the pulmonary artery arising mostly (>50%) from the left ventricle and with the aorta arising entirely from the right ventricle; accordingly cases with double outlet right ventricle were excluded.

CLINICAL FEATURES

The clinical data are summarised in Table 1. All six patients presented with signs of congestive heart failure and cyanosis; in three the brachial and femoral pulses were equal and normal in amplitude, while in the others the femoral pulses were either absent or very weak; differential cyanosis was never observed.

In the electrocardiogram two out of six showed isolated left ventricular hypertrophy (Fig. 1, case 2) and two had biventricular hypertrophy; in one there was right ventricular hypertrophy and in the sixth the electrocardiogram was normal, when first seen at 1 day of age.

The chest x-ray showed cardiothoracic ratio varying from 0.58 to 0.78 and increased pulmonary vascularity in all the patients. An M-mode echocardiogram was recorded in one instance (Fig. 2, case 3): the right ventricular dimension was reduced; there were two atrioventricular valves with continuity between the posterior one and the pulmonary artery; the anterior great artery (aorta) was decreased in diameter.

Three patients (cases 4 to 6) underwent cardiac catheterisation (Table 2). The oxygen saturation

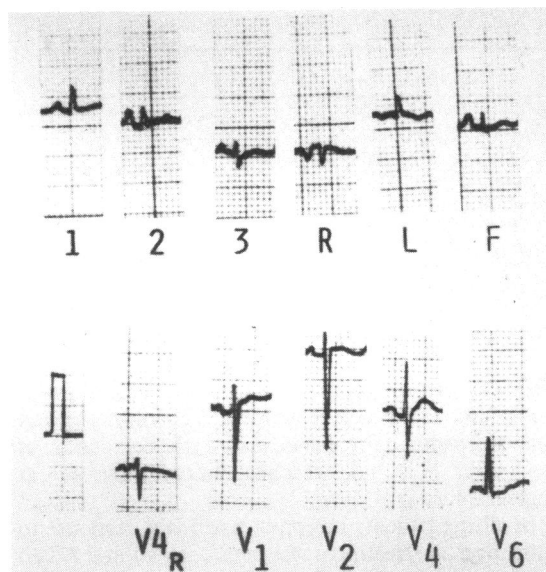


Fig. 1 Case 2. Electrocardiogram, showing a QRS axis of $+30^\circ$, left ventricular hypertrophy, and underdevelopment of the right ventricular electrical forces.

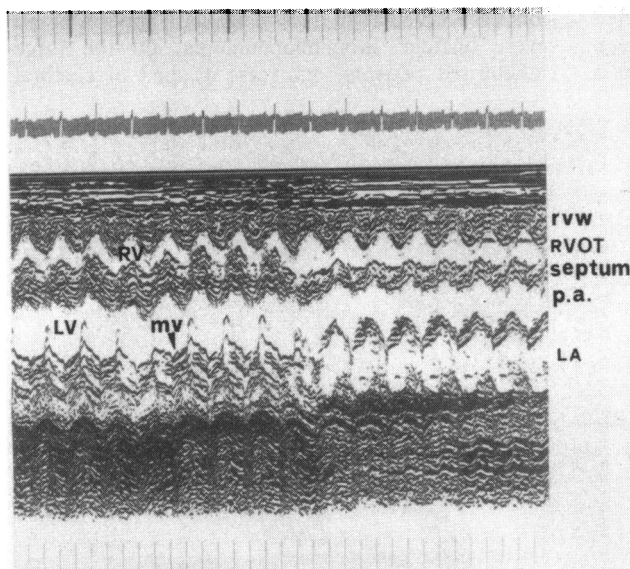


Fig. 2 Case 3. M-mode echocardiogram showing decreased right ventricular dimension and mitropulmonary continuity. The anterior wall of the pulmonary artery is entirely aligned with the interventricular septum.

Abbreviations: rvw, right ventricular wall; RVOT, right ventricular outflow tract; p.a., pulmonary artery; LA, left atrium; RV, right ventricle; LV, left ventricle; mv, mitral valve.

Table 1 Clinical data

Case No.	Sex	Age on admission	Cyanosis	CHF	Femoral pulses	Electrocardiogram		X-ray	
						LVH	RVH	C/T	Pulmonary vascular markings
1	M	1 d	1 ⁺	3 ⁺	Very weak	Yes	No	0.65	↑
2	M	6 d	1 ⁺	3 ⁺	Absent	Yes	No	0.78	↑
3	M	15 d	3 ⁺	2 ⁺	Normal	Yes	Yes	0.60	↑
4	M	55 d	1 ⁺	2 ⁺	Normal	No	Yes	0.60	↑
5	F	30 d	2 ⁺	3 ⁺	Normal	Yes	Yes	0.67	↑
6	M	1 d	2 ⁺	1 ⁺	Weak	No	No	0.58	↑
7*	M	5 mth	—	—	—	—	—	—	—

*Other clinical data not available.

Abbreviations: LVH, left ventricular hypertrophy; RVH, right ventricular hypertrophy; C/T, cardiothoracic ratio; CHF, congestive heart failure.

Table 2 Cardiac catheterisation data

Pressure (mmHg)									
Case No.	RA M	RV S/D	Ascending aorta S/D M	Descending aorta S/D M	LA M	LV S/D	PA S/D M	After Rashkind procedure	
								RA M	LA M
4	6	80/15	70/35 50		20	70/15	75/35 55	10	13
5	8	70/10	88/45 60	78/20 41	8	65/10	75/25 25		
6	3	80/4	80/40 60		5			0	0

showed a significant left-to-right shunt at the ventricular level in all of them. Pulmonary arterial hypertension was present in the two cases (cases 4 and 5) in which the pulmonary artery was entered; in only one case (case 5) was the descending aortic pressure obtained, showing a systolic gradient of 10 mmHg between ascending and descending aorta. In this case aortography showed a "stop" of the contrast medium at the aortic isthmus (Fig. 3a), which was interpreted as an atresia of the aortic arch, whereas the surgeon found a distinct coarctation. In all three patients a

diagnosis of transposition of the great arteries with ventricular septal defect was made by right and left cineventriculography; in one (case 4) the coarctation of the aorta was not recognised clinically or at angiography.

In all the patients but one (case 6) the response to anticongestive treatment was poor. Cases 1 and 2 died three days and a few hours, respectively, after admission; case 3 died at 40 days of age with superimposed severe bilateral bronchopneumonia. Of the three catheterised patients, one (case 4) died of cerebral

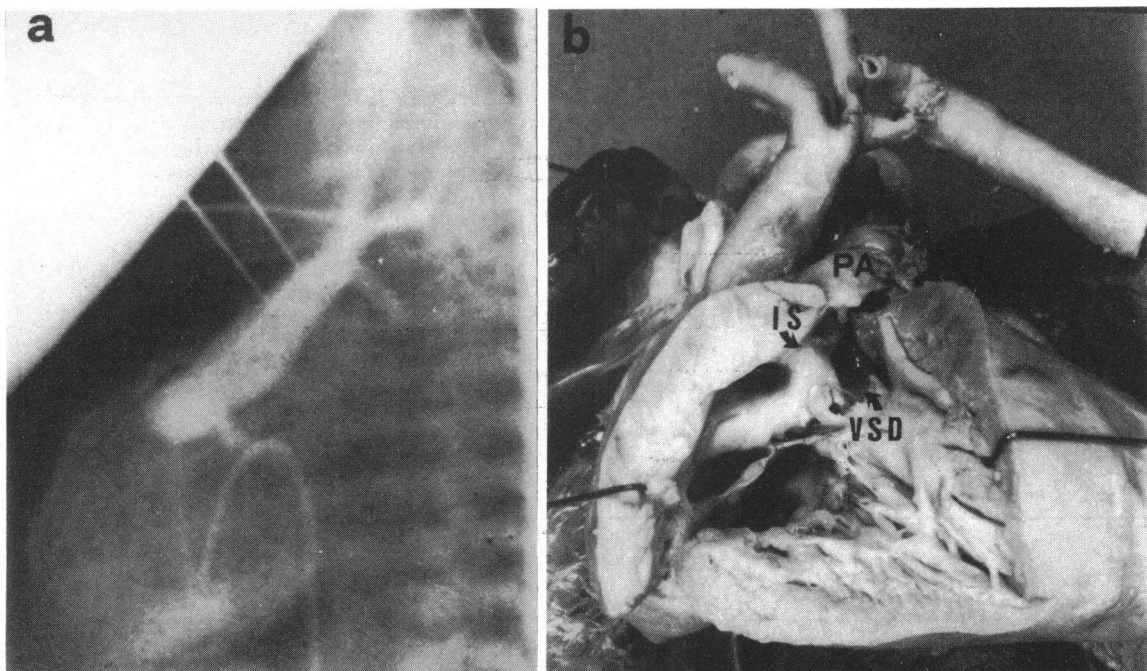


Fig. 3 Case 5. (a) Ascending aortography shows tubular hypoplasia of the aortic arch with complete interruption of the contrast medium at the level of the aortic isthmus. (b) The right ventricular view of the heart specimen discloses a subaortic infundibular stenosis caused by rightward displacement of the infundibular septum (IS), moderate dextroposition of the pulmonary artery (PA), and the subpulmonary ventricular septal defect (VSD). Note the pulmonary banding and the surgically attempted relief of the aortic arch obstruction.

O₂ Saturation (%)

SVC	RA	RV	Ascending aorta	Descending aorta	LA	LV	PA	PV	After Rashkind procedure	
									RV	Ascending aorta
53	55	72	68		91					
70	75	84		94	98	95	90	96		
20	18	29	36		83	75		90	72	

Abbreviations: RA, right atrium; RV, right ventricle; LA, left atrium; LV, left ventricle; PA, pulmonary artery; SVC, superior vena cava; M, mean pressure; S/D, systolic/diastolic pressure; PV, pulmonary vein.

haemorrhage five days after operation (persistent ductus arteriosus ligation plus pulmonary artery banding); another (case 5) died at 2 months of age, 48 hours after operation (resection of coarctation with end to end anastomosis of the aorta, pulmonary artery banding, and persistent ductus arteriosus ligation); the third (case 6) is the only patient who has survived and is now awaiting operation after a second cardiac catheterisation.

NECROPSY FINDINGS

The most relevant observations at necropsy are reported in Table 3. Visceroatrial situs solitus, laevocardia, atrioventricular concordance, and ventriculoarterial discordance were present in all the cases. The ascending aorta was small and the pulmonary artery large, the ratio between the diameters of the pulmonary artery and ascending aorta being more than 1.5:1 in three cases. Patent or closing ductus arteriosus

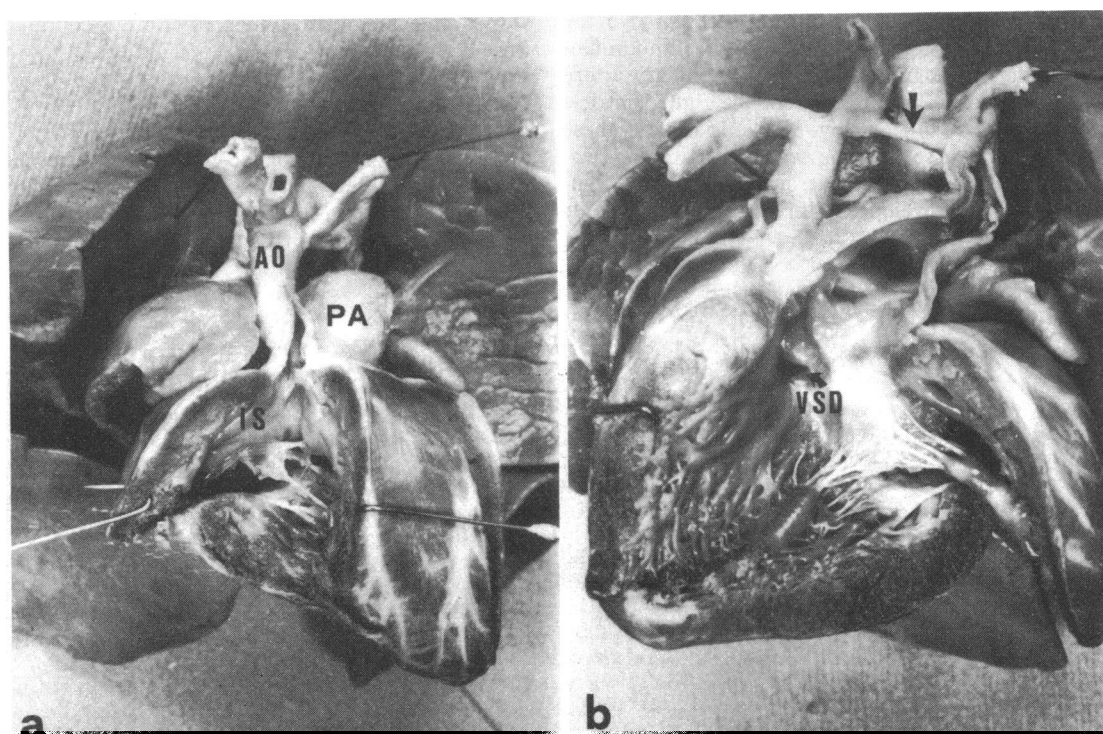


Fig. 4 Case 2. (a) The right ventricle has a hypoplastic trabecular component and an obstructed aortic outflow resulting from anterior deviation of the infundibular septum (IS). Note the extreme dilatation of the pulmonary artery (PA) when compared with the small ascending aorta (AO). (b) The view from the left ventricle shows the ventricular septal defect (VSD) caused by infundibulovenricular malalignment and mild dextroposition of the pulmonary artery. Note the atretic aortic arch (arrow) between the left carotid and left subclavian artery.

Table 3 Necropsy findings

Case No.	PA/Ao diameter ratio	Right ventricle Inflow	Trabecular portion	Outflow	VSD	% PA dextroposition	Aortic arch obstruction	PDA
1	1.7:1	Normal	Moderate hypoplasia	Normal	Subpulmonary malalignment	40%	Atresia type A*	Large
2	2:1	Normal	Severe hypoplasia	Subaortic stenosis	Subpulmonary malalignment	10%	Atresia type B*	Large
3	1.2:1	Normal	Normal	Normal	Subpulmonary malalignment	20%	Preductal coarctation	Closing
4	1.3:1	Normal	Moderate hypoplasia	Subaortic stenosis	Subpulmonary malalignment	15%	Tubular hypoplasia	Closing†
5	1.5:1	Normal	Normal	Subaortic stenosis	Subpulmonary malalignment	30%	Preductal†	Closing†
6‡								
7	2:1	Very stenotic	Severe hypoplasia	Moderate hypoplasia	Perimembranous VSD	—	Atresia type A*	Closing

*Celoria and Patton classification.⁶

†Surgical observation.

‡Case 6 is still alive.

Abbreviations: PA, pulmonary artery; Ao, ascending aorta; VSD, ventricular septal defect; PDA, persistent ductus arteriosus.

was observed in four cases, while in two it had been surgically ligated. The aortic arch appeared atretic in three instances, type A of Celoria and Patton⁶ in two and type B in one; preductal coarctation was present in two and tubular hypoplasia in one. All specimens had an associated ventricular septal defect. In five the defect was subpulmonary because of infundibulo-ventricular malalignment and the pulmonary artery was dextroposed, its origin from the right ventricle varying from 10 to 40% (Fig. 3b, 4). In three of these the infundibular septum showed an anterior displacement, creating subaortic narrowing. The right ventricle was underdeveloped in three hearts, mostly in its trabecular component.

In the sixth specimen, instead, the ventricular septal defect was small, perimembranous, and the pulmonary artery arose entirely from the left ventricle. The tricuspid valve appeared very stenotic and the right ventricle extremely hypoplastic (Fig. 5).

In all the cases mitro-pulmonary fibrous continuity was present.

Discussion

Complete transposition of the great arteries is usually classified into three physiological clinical entities: (1) simple transposition of the great arteries with intact ventricular septum characterised by conspicuous

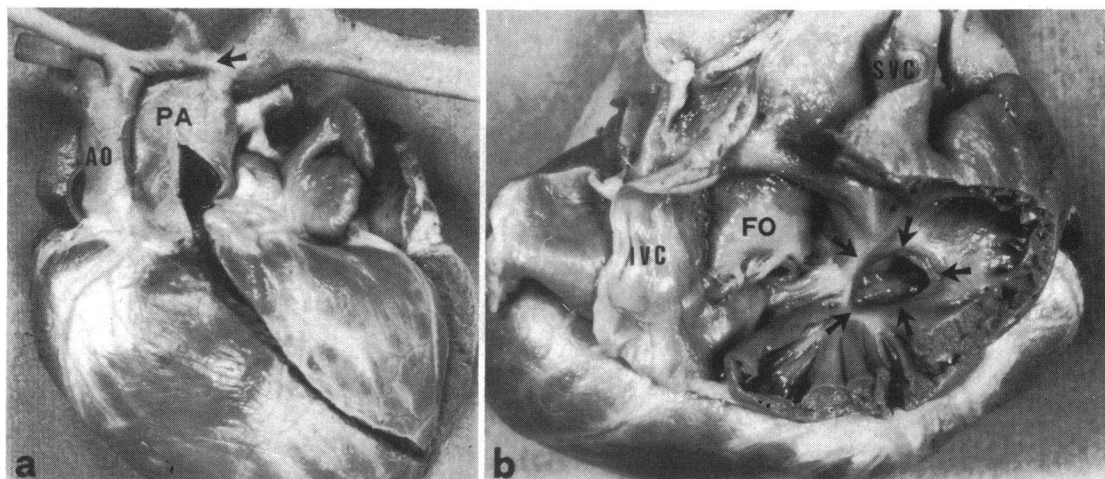


Fig. 5 Case 6. (a) External view of the heart showing severe hypoplasia of the aortic arch with preductal coarctation (arrow), hypoplastic ascending aorta (AO), and dilated pulmonary artery (PA). (b) View of the opened right atrium showing that the tricuspid orifice is very small (arrows) compared with the fossa ovalis (FO). The foramen ovale is restrictive and the superior (SVC) and inferior (IVC) vena cavae are dilated.

hypoxia, caused by lack of mixing between pulmonary and systemic circulation; (2) transposition of the great arteries with large ventricular septal defect in which signs of congestive heart failure predominate in the clinical picture; and (3) transposition of the great arteries with ventricular septal defect and pulmonary stenosis in which cyanosis is prominent, caused by decreased pulmonary blood flow, and the clinical findings parallel those in infants with tetralogy of Fallot.⁷

The syndrome of coarctation of the aorta in transposition of the great arteries has been considered extremely rare⁸ and, accordingly, has never received sufficient attention. Our experience, together with other recent reports,⁹⁻¹⁰ suggest that aortic arch anomalies are not so rare and the clinical manifestations and the pathological findings of this association are sufficiently distinctive as to be considered a separate nosographic entity among transposition of the great arteries.

Besides the presence of transposition of the great arteries and obstruction of the aortic arch, the following morphological features delineate this anatomical complex: (a) ventricular septal defect, always present, mostly subpulmonary and associated with dextroposition of the pulmonary artery; (b) mild to moderate right ventricular underdevelopment mainly of the trabecular component; and (c) right ventricular inflow, rarely, or outflow, more frequently, obstruction (caused by tricuspid stenosis or infundibular subaortic stenosis, respectively). Usually, the lower part of the systemic circulation is ductus-dependent.

From the clinical standpoint, cyanosis, early and severe congestive heart failure, and decreased amplitude of the femoral pulses characterise most of these patients. This picture of coarctation of the aorta in transposition of the great arteries is often associated with the presence of left ventricular hypertrophy on the electrocardiogram, either isolated (2/6) or combined with right ventricular hypertrophy (2/6). It is noteworthy that isolated left ventricular hypertrophy was observed just in those two cases with an atric aortic arch and moderate to severe right ventricular hypoplasia. The decreased right ventricular electrical forces and the pressure and volume overload of the left ventricle account for this paradoxical electrocardiographic pattern in transposition of the great arteries.

Anterior displacement of the infundibular septum, as seen in three of our cases, has been suggested as a cause of subaortic stenosis in transposition of the great arteries, and obstruction of the aortic arch has also been described.⁵⁻⁹⁻¹⁰ Unequal partitioning of the blood flow between the aorta and pulmonary artery in the presence of a ventricular septal defect may be advocated as the fetal pathogenetic mechanism for the

underdevelopment of the aortic arch, as postulated for aortic arch anomalies in normally related great arteries.¹¹ Preferential blood streaming toward the pulmonary artery and ductus arteriosus may also be the result of inlet obstruction of the right ventricle, as in our case 7, or of dextroposition of the pulmonary artery, as in double outlet right ventricle with subpulmonary ventricular septal defect (Taussig-Bing), which is often associated with coarctation or interruption of the aorta.

In conclusion, we believe that, among transpositions of the great arteries, the form with aortic arch obstruction merits being viewed as a distinct physiological clinical type because it is relatively frequent and its pathological anatomy is distinctive.

This type of transposition is unfortunately associated with a worse clinical and surgical prognosis than is now the case in the other three major types.

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